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CORRECTION

Correction to: Multiple endocrine neoplasia type 1: analysis of germline *MEN1* mutations in the Italian multicenter MEN1 patient database

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The original version of this article unfortunately contained a mistake in Table 2. The Table 2 was truncated in the original publication. The full Table 2 is given below.
The original article has been corrected.

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Table 2 Main characteristics of *MEN1* mutations in our MEN1 patients

Mutation	References	Type	Exon	Intron	Affected codon	Amino acid substitution	Premature stop codon	Main effect on menin protein	Number of MEN1 families bearing the mutation (total members)	Number of non-familial MEN1 cases bearing the mutation	Total number of MEN1 patients bearing the mutation
g.197delA	Novel[b]	Frameshift (deletion)	2	NA	29	NA	118 (TGA)	Shortened menin of only 117 amino acids, lacking all three NLSs	1 (2)	0	2
g.301_305dup5	Bassett [17]	Frameshift (insertion)	2	NA	67	NA	120 (TGA)	Shortened menin of only 119 amino acids, lacking all three NLSs	0	1	1
g.302 insA	Concolino [3]	Frameshift (insertion)	2	NA	64	NA	116 (TGA)	Shortened menin of only 115 amino acids, lacking all three NLSs	1 (4)	0	4
g.308_312ins5	Giraud [18]	Frameshift (insertion)	2	NA	66	NA	114 (TGA)	Shortened menin of only 113 amino acids, lacking all three NLSs	2 (5)	0	5
g.317_320ins4	Vannucci [19]	Frameshift (insertion)	2	NA	69	NA	117 (TGA)	Shortened menin of only 116 amino acids, lacking all three NLSs	1 (6)	0	6
g.317_321ins5	Cebrian [20]	Frameshift (insertion)	2	NA	69	NA	120 (TGA)	Shortened menin of only 119 amino acids, lacking all three NLSs	3 (7)	1	8
g.317delC	Agarwal [21]	Frameshift (deletion)	2	NA	69	NA	118 (TGA)	Shortened menin of only 117 amino acids, lacking all three NLSs	2 (8)	0	8
g.335delA	Novel[b]	Frameshift (deletion)	2	NA	76	NA	118 (TGA)	Shortened menin of only 117 amino acids, lacking all three NLSs	0	1	1
g.357_360del4	Agarwal [21]	Frameshift (deletion)	2	NA	83	NA	118 (TGA)	Shortened menin of only 116 amino acids, lacking all three NLSs	0	1	1
g.359_362del4	Sakurai [22]	Frameshift (deletion)	2	NA	83	NA	117 (TGA)	Shortened menin of only 116 amino acids, lacking all three NLSs	4 (10)	0	10
g.445insC	Novel[b]	Frameshift (insertion)	2	NA	111	NA	116 (TGA)	Shortened menin of only 115 amino acids, lacking all three NLSs	0	1	1
g.531delC	Novel[b]	Frameshift (deletion)	2	NA	140	NA	184 (TAG)	Shortened menin of only 183 amino acids, lacking all three NLSs	1 (1)	0	1
g.579delG	Nuzzo [23]	Frameshift (deletion)	3	NA	157	NA	184 (TAG)	Shortened menin of only 183 amino acids, lacking all three NLSs	1 (4)	0	4
g.613delT	Novel[b]	Frameshift (deletion)	3	NA	167	NA	184 (TAG)	Shortened menin of only 183 amino acids, lacking all three NLSs	1 (3)	0	3
g.734_737del4	Novel[b]	Frameshift (deletion)	3	NA	208	NA	222 (TGA)	Shortened menin of only 221 amino acids, lacking all three NLSs	1 (5)	0	5
g.738_741del4	Chandrasekharappa [24]	Frameshift (deletion)	3	NA	210	NA	222 (TGA)	Shortened menin of only 221 amino acids, lacking all three NLSs	2 (4)	0	4
g.868delC	Novel[b]	Frameshift (deletion)	4	NA	253	NA	279 (TAG)	Shortened menin of only 278 amino acids, lacking all three NLSs	1 (1)	1	2
g.953_954delGA	Novel[b]	Frameshift (deletion)	6	NA	281	NA	315 (TGA)	Shortened menin of only 314 amino acids, lacking all three NLSs	1 (3)	0	3
g.1005delC	Ellard [25]	Frameshift (deletion)	6	NA	299	NA	367 (TAG)	Shortened menin of only 366 amino acids, lacking all the three NLSs	0	1	1
g.1059delC	Morelli [26]	Frameshift (deletion)	7	NA	317	NA	367 (TAG)	Shortened menin of only 366 amino acids, lacking all three NLSs	0	1	1
g.1060insC	Novel[b]	Frameshift (insertion)	7	NA	317	NA	368 (TAG)	Shortened menin of only 367 amino acids, lacking all three NLSs	0	1	1
g.1061delC	Novel[b]	Frameshift (deletion)	7	NA	317	NA	367 (TAG)	Shortened menin of only 366 amino acids, lacking all three NLSs	1 (3)	0	3

Table 2 (continued)

Mutation	References	Type	Exon	Intron	Affected codon	Amino acid substitution	Premature stop codon	Main effect on menin protein	Number of MEN1 families bearing the mutation (total members)	Number of non-familial MEN1 cases bearing the mutation	Total number of MEN1 patients bearing the mutation
g.1071delT	Morelli [26]	Frameshift (deletion)	7	NA	321	NA	367 (TAG)	Shortened menin of only 366 amino acids, lacking all three NLSs	1 (2)	0	2
g.1181delC[a]	Vannucci [19]	Frameshift (deletion)	8	NA	357	NA	367 (TAG)	Shortened menin of only 366 amino acids, lacking all three NLSs	1 (5)	0	5
g.1264delC	Morelli [26]	Frameshift (deletion)	8	NA	385	NA	444 (TAG)	Shortened menin of only 443 amino acids, lacking all three NLSs	1 (7)	0	7
g.1284delG	Agarwal [21]	Frameshift (deletion)	8	NA	392	NA	444 (TAG)	Shortened menin of only 443 amino acids, lacking all three NLSs	1 (1)	1	2
g.1364delC	Hai [27]	Frameshift (deletion)	9	NA	418	NA	444 (TAG)	Shortened menin of only 443 amino acids, lacking all three NLSs	1 (5)	0	5
g.1434delC	Novel[b]	Frameshift (deletion)	9	NA	441	NA	444 (TAG)	Shortened menin of only 443 amino acids, lacking all three NLSs	0	1	1
g.1449_1459del11	Giraud [18]	Frameshift (deletion)	9	NA	447	NA	526 (TGA)	Shortened menin of only 525 amino acids, lacking NLSa (AA 546–572) and NLS2 (AA 588–608) and with NLS1 (AA 479–497) altered because of aminoacidic changes from codon 447	1 (24)	0	24
g.1528_1534del7	Novel[b]	Frameshift (deletion)	10	NA	473	NA	530 (TGA)	Shortened menin of only 529 amino acids, lacking NLSa (AA 546–572) and NLS2 (AA 588–608) and with NLS1 (AA 479–497) altered because of aminoacidic changes from codon 473	1 (2)	0	2
g.1555insG	Morelli [26]	Frameshift (insertion)	10	NA	481	NA	530 (TGA)	Shortened menin of only 529 amino acids, lacking NLSa (AA 546–572) and NLS2 (AA 588–608) and with NLS1 (AA 479–497) altered because of aminoacidic changes from codon 481	3 (9)	0	9
g.1571delC	Novel[b]	Frameshift (deletion)	10	NA	487	NA	529 (TGA)	Shortened menin of only 528 amino acids, lacking NLSa (AA 546–572) and NLS2 (AA 588–608) and with NLS1 (AA 479–497) altered because of aminoacidic changes from codon 487	1 (2)	0	2
g.1631delG	Novel[b]	Frameshift (deletion)	10	NA	507	NA	558 (TGA)	Shortened menin of only 557 amino acids, lacking NLSa (AA 546–572) and NLS2 (AA 588–608). NLS1 (AA 479–497) remains intact.	1 (2)	0	2
g.1656insC	Agarwal [21]	Frameshift (insertion)	10	NA	516	NA	530 (TGA)	Shortened menin of only 529 amino acids, lacking NLSa (AA 546–572) and NLS2 (AA 588–608). NLS1 (AA 479–497) remains intact.	9 (25)	2	27
g.1659insG	Bartsch [28]	Frameshift (insertion)	10	NA	517	NA	530 (TGA)	Shortened menin of only 529 amino acids, lacking NLSa (AA 546–572) and NLS2 (AA 588–608). NLS1 (AA 479–497) remains intact.	1 (2)	0	2
g.1671_1680del11	Concolino [3]	Frameshift (deletion)	10	NA	521	NA	526 (TGA)	Shortened menin of only 525 amino acids, lacking NLSa (AA 546–572) and NLS2 (AA 588–608). NLS1 (AA 479–497) remains intact.	3 (7)	0	7
g.1690delG	Novel[b]	Frameshift (deletion)	10	NA	527	NA	561 (TGA)	Shortened menin of only 560 amino acids lacking part of NLSa (AA 546–572) and all NLS2 (AA 588–608). NLS1 (AA 479–497) remains intact.	1 (1)	0	1
g.1705_1706insGG	Novel[b]	Frameshift (insertion)	10	NA	532	NA	561 (TGA)	Shortened menin of only 560 amino acids, lacking part of NLSa (AA 546–572) and all NLS2 (AA 588–608). NLS1 (AA 479–497) remains intact.	1 (3)	0	3

Table 2 (continued)

Mutation	References	Type	Exon	Intron	Affected codon	Amino acid substitution	Premature stop codon	Main effect on menin protein	Number of MEN1 families bearing the mutation (total members)	Number of non-familial MEN1 cases bearing the mutation	Total number of MEN1 patients bearing the mutation
g.1757insA	Novel[b]	Frameshift (insertion)	10	NA	549	NA	556 (TGA)	Shortened menin of only 555 amino acids, lacking part of NLSa (AA 546–572) and all NLS2 (AA 588–608). NLS1 (AA 479–497) remains intact.	1 (1)	0	1
g.1786delA	Asteria [29]	Frameshift (deletion)	10	NA	559	NA	560 (TGA)	Shortened menin of only 559 amino acids, lacking part of NLSa (AA 546–572) and all NLS2 (AA 588–608). NLS1 (AA 479–497) remains intact.	1 (5)	0	5
g.1937insC	Novel[b]	Frameshift (insertion)	10	NA	609	Stop611Leu	Loss of stop codon (TGA) at position 611	Insertion of a cytosine after codon 609 that leads to the loss of the stop codon at position 611. A novel codon stop (TGA) is generated at codon 632, presumably leading to a menin protein longer than normal (631 amino acids)	1 (3)	0	3
g.468_470del3	Agarwal [21]	In-frame deletion	2	NA	120	Lys120del	NA	Loss of Lysine at position 120, affecting binding site of SMAD3 (aa 40–278) and NM23H1 (aa 1–486). Menin protein of 609 amino acids.	0	1	1
g.674_691del18	Giacché [30]	In-frame deletion	3	NA	189–194	Loss of amino acids 189–194	NA	Loss of amino acids at position 189–194, affecting binding sites of JUND (aa 39–242), NMHCII-A (aa 154–306), SMAD3 (aa 40–278), HDAC1 (aa 145–450) and NM23H1 (aa 1–486). Menin protein of 604 amino acids.	0	1	1
g.908_910del3	Papi [31]	In-frame deletion	5	NA	266–267	Leu267loss	NA	Loss of Leucine at position 267, affecting binding sites of NMHCII-A (aa 154–306), FANCD2 (aa 219–395), SMAD3 (aa 40–278), HDAC1 (aa 145–450) and NM23H1 (aa 1–486). Menin protein of 609 amino acids.	1 (3)	0	3
g.1433_1438del6	Novel[b]	In-frame deletion	9	NA	441–443	Loss of Gln442 and Ser443	NA	Loss of Glutamine at position 442 and Serine at position 443, affecting binding sites of NM23H1 (aa 1–486), RPA2 (aa 286–448) and CHES1 (aa 428–610). Menin protein of 608 amino acids.	1(2)	0	2
Gln64Stop	Langer [32]	Nonsense	2	NA	64 (CAG > TAG)	NA	64 (TAG)	Shortened menin of only 63 amino acids, lacking all three NLSs	1 (3)	0	3
Trp126Stop	Bassett [17]	Nonsense	2	NA	126 (TGG > TAG)	NA	126 (TAG)	Shortened menin of only 125 amino acids, lacking all three NLSs	2 (4)	0	4
Glu274Stop	Novel[b]	Nonsense	5	NA	274 (GAA > TAA)	NA	274 (TAA)	Shortened menin of only 273 amino acids, lacking all three NLSs	1 (1)	0	1
Thr341Stop	Cebrian [33]	Nonsense	7	NA	341 (TGG > TGA)	NA	341 (TGA)	Shortened menin of only 340 amino acids, lacking all three NLSs	1 (3)	0	3
Arg415Stop	Lemmens [34]	Nonsense	9	NA	415 (CGA > TGA)	NA	415 (TGA)	Shortened menin of only 414 amino acids, lacking all three NLSs	7 (33)	2	35
Gln442Stop	Shimizu [35]	Nonsense	9	NA	442 (CAG > TAG)	NA	442 (TAG)	Shortened menin of only 441 amino acids, lacking all three NLSs	2 (3)	0	3

Table 2 (continued)

Mutation	References	Type	Exon	Intron	Affected codon	Amino acid substitution	Premature stop codon	Main effect on menin protein	Number of MEN1 families bearing the mutation (total members)	Number of non-familial MEN1 cases bearing the mutation	Total number of MEN1 patients bearing the mutation
Gln450Stop	Hai [36]	Nonsense	9	NA	450 (CAG > TAG)	NA	450 (TAG)	Shortened menin of only 449 amino acids, lacking all three NLSs	2 (4)	2	6
Arg460Stop	Agarwal [21]	Nonsense	10	NA	460 (CGA > TGA)	NA	460 (TGA)	Shortened menin of only 459 amino acids, lacking all three NLSs	1 (13)	2	15
Glu474Stop	Corbetta [37]	Nonsense	10	NA	474 (GAA > TAA)	NA	474 (TAA)	Shortened menin of only 473 amino acids, lacking all three NLSs	0	1	1
Gly508Stop	Morelli [26]	Nonsense	10	NA	508 (CAG > TAG)	NA	508 (TAG)	Shortened menin of only 507 amino acids, lacking NLSa (aa 546–572) and NLS2 (aa 588–608). NLS1 remains intact	1 (2)	0	2
Arg527Stop	Chandrasekharappa [24]	Nonsense	10	NA	527 (CGA > TGA)	NA	527 (TGA)	Shortened menin of only 526 amino acids, lacking NLSa (aa 546–572) and NLS2 (aa 588–608). NLS1 remains intact	1 (2)	0	2
Glu556Stop	Jap [38]	Nonsense	10	NA	556 (GAG > TAG)	NA	556 (TAG)	Shortened menin of only 555 amino acids, lacking part of NLSa (aa 546–572) and the entire NLS2 (aa 588–608). NLS1 remains intact	0	1	1
Lys557Stop	Novel[b]	Nonsense	10	NA	557 (AAG > TAG)	NA	557 (TAG)	Shortened menin of only 556 amino acids, lacking part of NLSa (aa 546–572) and the entire NLS2 (aa 588–608). NLS1 remains intact	2 (4)	0	4
Met1Val	Villablanca [39]	Missense	2	NA	1 (ATG > GTG)	Met > Val	NA	Initial Metionin at codon 1 is substituted by a Valine. Usually initial Metionine is enzimatically removed at post-traduction level. The presence of a Valine at position 1 affects binding sites of JUND (aa 1–40), NM23H1 (aa 1–486) and RPA2 (aa 1–40). Menin protein of 611 amino acids	1 (1)	0	1
Glu45Gln	Griniatsos [40]	Missense	2	NA	45 (GAG > CAG)	Glu > Gln	NA	Single amino acid substitution at position 45, affecting binding sites of NM23H1 (aa 1–486) and SMAD3 (aa 40–278)	2 (7)	0	7
Glu45Lys	Morelli [26]	Missense	2	NA	45 (GAG > AAG)	Glu > Lys	NA	Single amino acid substitution at position 45, affecting binding site of NM23H1 (aa 1–486) and SMAD3 (aa 40–278)	0	1	1
Arg137Trp	Novel[b]	Missense	2	NA	137 (CGG > TGG)	Arg > Trp	NA	Single amino acid substitution at position 137, affecting binding sites of NM23H1 (aa 1–486) and SMAD3 (aa 40–278)	1 (2)	0	2
Phe146Ser	Vannucci [19]	Missense	2	NA	146 (TTC > TCC)	Phe > Ser	NA	Single amino acid substitution at position 146, affecting binding sites of JUND (aa 139–242), NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	1 (2)	0	2
Asp153Glu	Filopanti [41]	Missense	3	NA	153 (GAC > GAG)	Asp > Glu	NA	Single amino acid substitution at position 153, affecting binding sites of JUND (aa 139–242), NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	4 (8)	0	8

Table 2 (continued)

Mutation	References	Type	Exon	Intron	Affected codon	Amino acid substitution	Premature stop codon	Main effect on menin protein	Number of MEN1 families bearing the mutation (total members)	Number of non-familial MEN1 cases bearing the mutation	Total number of MEN1 patients bearing the mutation
Gly163Arg	Novel[b]	Missense	3	NA	163 (GGG > AGG)	Gly > Arg	NA	Single amino acid substitution at position 163, affecting binding sites of JUND (aa 139–242), NMHCII-A (aa 154–306) NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	1 (1)	0	1
His181Asp	Novel[b]	Missense	3	NA	181 (CAT > GAT)	His > Asp	NA	Single amino acid substitution at position 181, affecting binding sites of JUND (aa 139–242), NMHCII-A (aa 154–306) NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	0	1	1
Val196Gly	Vannucci [19]	Missense	3	NA	196 (GTC > GGC)	Val > Gly	NA	Single amino acid substitution at position 196, affecting binding sites of JUND (aa 139–242), NMHCII-A (aa 154–306) NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	1 (7)	0	7
His199Asp	Novel[b]	Missense	3	NA	199 (CAC > GAC)	His > Asp	NA	Single amino acid substitution at position 199, affecting binding sites of JUND (aa 139–242), NMHCII-A (aa 154–306) NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	0	1	1
Val215Met	Morelli [26]	Missense	3	NA	215 (GTG > ATG)	Val > Met	NA	Single amino acid substitution at position 215, affecting binding sites of JUND (aa 139–242), NMHCII-A (aa 154–306) NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	1 (2)	0	2
Trp220Arg	Novel[b]	Missense	4	NA	220 (TGG > CGG)	Trp > Arg	NA	Single amino acid substitution at position 220, affecting binding sites of JUND (aa 139–242), FANCD2 (aa 219–395), NMHCII-A (aa 154–306) NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	1 (2)	1	3
Leu249Pro[a]	Vannucci [19]	Missense	4	NA	249 (CTG > CCG)	Leu > Pro	NA	Single amino acid substitution at position 249, affecting binding sites of NM23H1 (aa 1–486), FANCD2 (aa 219–395), NMHCII-A (aa 154–306), SMAD3 (aa 40–278) e HDAC1 (aa 145–450).	1 (5)	0	5
Leu256Phe	Tham [42]	Missense	4	NA	256 (CTT > TTT)	Leu > Phe	NA	Single amino acid substitution at position 256, affecting binding sites of FANCD2 (aa 219–395), NMHCII-A (aa 154–306) NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	0	1	1
Leu273Pro	Novel[b]	Missense	5	NA	273 (CTG > CCG)	Leu > Pro	NA	Single amino acid substitution at position 273, affecting binding sites of FANCD2 (aa 219–395), NMHCII-A (aa 154–306) NM23H1 (aa 1–486), SMAD3 (aa 40–278) and HDAC1 (aa 145–450)	1 (6)	0	6
Ala284Val	Novel[b]	Missense	6	NA	284 (GCA > GTA)	Ala > Val	NA	Single amino acid substitution at position 284, affecting binding sites of FANCD2 (aa 219–395), NMHCII-A (aa 154–306) NM23H1 (aa 1–486) and HDAC1 (aa 145–450)	1 (3)	0	3
Thr344Arg	Agarwal [21]	Missense	7	NA	344 (ACG > AGG)	Thr > Arg	NA	Single amino acid substitution at position 344, affecting binding sites of JUND (aa 323–428) and NF-kB (aa 305–381), FANCD2 (aa 219–395), RPA2 (aa 286–448), NM23H1 (aa 1–486) and HDAC1 (aa 145–450)	3 (8)	1	9

Table 2 (continued)

Mutation	References	Type	Exon	Intron	Affected codon	Amino acid substitution	Premature stop codon	Main effect on menin protein	Number of MEN1 families bearing the mutation (total members)	Number of non-familial MEN1 cases bearing the mutation	Total number of MEN1 patients bearing the mutation
Cys354Phe	Vannucci [19]	Missense	8	NA	354 (TGC > TTC)	Cys > Phe	NA	Single amino acid substitution at position 354, affecting binding sites of JUND (aa 323–428) and NF-kB (aa 305–381), FANCD2 (aa 219–395), RPA2 (aa 286–448), NM23H1 (aa 1–486) and HDAC1 (aa 145–450)	1 (4)	0	4
Pro390Arg	Novel[b]	Missense	8	NA	390 (CCG > CGG)	Pro > Arg	NA	Single amino acid substitution at position 354, affecting binding sites of JUND (aa 323–428), mSin3A (aa 371–387), FANCD2 (aa 219–395), RPA2 (aa 286–448), NM23H1 (aa 1–486) and HDAC1 (aa 145–450)	1 (2)	0	2
Leu413Pro	Asteria [29]	Missense	9	NA	413 (CTG > CCG)	Leu > Pro	NA	Single amino acid substitution at position 413, affecting binding sites of JUND (323–428), NM23H1 (1–486), RPA2 (286–448) and HDAC1 (aa 145–450)	0	1	1
Leu413Arg	Toledo [43]	Missense	9	NA	413 (CTG > CGG)	Leu > Arg	NA	Single amino acid substitution at position 413, affecting binding sites of JUND (323–428), NM23H1 (1–486), RPA2 (286–448) and HDAC1 (aa 145–450)	0	2	2
Asp418Asn	Bassett [17]	Missense	9	NA	418 (GAC > AAC)	Asp > Asn	NA	Single amino acid substitution at position 418, affecting binding sites of JUND (323–428), NM23H1 (1–486), RPA2 (286–448) and HDAC1 (aa 145–450)	2 (3)	1	4
Gly419Val	Novel[b]	Missense	9	NA	419 (GGC > GTC)	Gly > Val	NA	Single amino acid substitution at position 419, affecting binding sites of JUND (aa 323–428), NM23H1 (aa 1–486), RPA2 (286–448) and HDAC1 (aa 145–450)	0	1	1
Trp423Arg	Cebrian [20]	Missense	9	NA	423 (TGG > CGG)	Trp > Arg	NA	Single amino acid substitution at position 423, affecting binding sites of JUND (aa 323–428), NM23H1 (aa 1–486), RPA2 (aa 286–448) and HDAC1 (aa 145–450)	1 (3)	0	3
Leu444Pro	Cetani [44]	Missense	9	NA	444 (CTA > CCA)	Leu > Pro	NA	Single amino acid substitution at position 444, affecting binding sites of NM23H1 (aa 1–486), RPA2 (aa 286–448), HDAC1 (aa 145–450) and CHES1 (aa 428–610)	4 (11)	1	12
Phe447Ser	Agarwal [21]	Missense	9	NA	447 (TTT > TCT)	Phe > Ser	NA	Single amino acid substitution at position 447, affecting binding sites of NM23H1 (aa 1–486), RPA2 (aa 286–448), HDAC1 (aa 145–450) and CHES1 (aa 428–610)	1 (1)	0	1
g.104 G > A	Novel[b]	Splicing-site	2	NA	NA	NA	NA	Altering the splicing site 6 bases before the initial ATG codon within the exon 1, maybe altering binding sites of JUND (aa 1–40), NM23H1 (aa 1–486) and RPA2 (aa 1–40)	1 (3)	0	3
g.556-3 C > G	Burgess [45]	Splicing-site	NA	2	NA	NA	NA	Affecting the splicing site between intron 2 and exon 3	0	1	1
g.765-1 G > C	Balogh [46]	Splicing-site	NA	3	NA	NA	NA	Affecting the splicing site between intron 3 and exon 4	1 (4)	0	4
g.765-1 G > A	Novel[b]	Splicing-site	NA	3	NA	NA	NA	Affecting the splicing site between intron 3 and exon 4	0	1	1

Table 2 (continued)

Mutation	References	Type	Exon	Intron	Affected codon	Amino acid substitution	Premature stop codon	Main effect on menin protein	Number of MEN1 families bearing the mutation (total members)	Number of non-familial MEN1 cases bearing the mutation	Total number of MEN1 patients bearing the mutation
g.893 + 1 G > A	Morelli [26]	Splicing-site	NA	4	NA	NA	NA	Affecting the splicing site between exon 4 and intron 4	1 (7)	0	7
g.893 + 1 G > C	Poncin [47]	Splicing-site	NA	4	NA	NA	NA	Affecting the splicing site between exon 4 and intron 4	1 (3)	0	3
g.894-9 G > A	Gortz [48]	Splicing-site	NA	4	NA	NA	NA	Affecting the splicing site between intron 4 and exon 5	5 (17)	1	18
g.935-2 A > G	Toliat [49]	Splicing-site	NA	5	NA	NA	NA	Affecting the splicing site between intron 5 and exon 6	1 (2)	0	2
g.935-18 delGA	Novel[b]	Splicing-site	NA	5	NA	NA	NA	Affecting the splicing site between intron 5 and exon 6	0	1	1
g.1159 + 1 G > A	Bassett [17]	Splicing-site	NA	7	NA	NA	NA	Affecting the splicing site between exon 7 and intron 7	1 (2)	1	3
g.1159 + 2 T > C	Han [50]	Splicing-site	NA	7	NA	NA	NA	Affecting the splicing site between exon 7 and intron 7	1 (1)	0	1
NA	NA	Large intra-genic deletion	Entire exon 1 and part of exon 2	1	NA	NA	NA	Loss of the entire exon 1 and part of exon 2. Loss of binding sites with JUND (aa 1–40), RPA2 (aa 1–40), NM23H1 (aa 1–486), SMAD3 (aa 40–278).	1 (2)	0	2
NA	NA	Large intra-genic deletion	1, 2 and 3	1 and 2	NA	NA	NA	Loss of exons 1, 2 and 3. Loss of binding sites with JUND (aa 1–40), NM23H1 (aa 1–486), NMHCII-A (aa 154–306), SMAD3 (aa 40–278) and HDAC1 (aa 145–450	1 (3)	0	3
NA	NA	Large multiple intra-genic deletions	Large intragenic deletions along the entire gene	NA	NA	NA	NA	Large intragenic deletions along the entire gene	2 (4)	0	4

NA non-applicable. *g.* genomic (nucleotide counting has been considered from the first nucleotide of the exon 1, excluding introns). Mutations indicated by

[a]Were both found in one family (bearing these two mutations on the same *MEN1* allele, inherited by the MEN1 index case from her paternal line)

[b]Mutations are indicated as "novel" if they are not reported in the Human Mutation Database (http://www.hgmd.cf.ac.uk/ac/index.php) or they have never been published before this study